CONTENTS

No 1 *FEBRUARY* 1985

Editorial .. 1

Discordant sex in one of three monozygotic triplets B DALLAPICCOLA, C STOMEO, G FERRANTI, A DI LECCI, AND M PURPURA .. 6

The estimation of recurrence risks in monogenic disorders using flanking marker loci R M WINTER .. 12

Investigation of human chromosome polymorphisms by scanning electron microscopy C J HARRISON, E M JACK, T D ALLEN, AND R HARRIS .. 16

The prevalence of translocations in parents of children with regular trisomy 21: a possible inter-chromosomal effect? R H LINDENBAUM, M HULTÉN, A McDERMOTT, AND M SEABRIGHT .. 24

Reduced penetrance in tuberous sclerosis M BARAITSER AND M A PATTON .. 29

Increased frequency of lymphocytic mitotic non-disjunction in recurrent spontaneous aborters R C JUBERG, J KNOPS, AND P N MOWREY .. 32

Profound limb deficiency, thoracic dystrophy, unusual facies, and normal intelligence: a new syndrome S A AL-AWADI, A S TEEBI, T I FARAG, K M NAGUIB, AND M Y EL-KHALIFA .. 36

Trigonocephaly and the Opitz C syndrome C SARGENT, J BURN, M BARAITSER, AND M E PEMBREY .. 39

Short rib-polydactyly syndrome: a single or heterogeneous entity? A re-evaluation prompted by four new cases R BERNSTEIN, J ISDALE, M PINTO, J DU TOIT ZAAIJMAN, AND T JENKINS .. 46

β^+-thalassaemia in the Po river delta region (northern Italy): genotype and β globin synthesis L DEL SENNO, M PIRASTU, R BARBIERI, F BERNARDI, D BUZZONI, G MARCHETTI, C PERROTTA, C VULLO, Y W KAN, AND F CONCONI .. 54

A family with three independent autosomal translocations associated with 7q32→7qter syndrome H N BASS, R S SPARKES, M M LESSNER, M FOX, B PHOENIX, AND J BERNAR .. 59

Case reports

De novo tandem duplication 9p (p12→p24) with normal GALT activity in red cells T MOTEGI, K WATANABE, N NAKAMURA, T HASEGAWA, AND Y YANAGAWA .. 64

A complex balanced rearrangement involving four chromosomes in an azoospermic man M T RODRIGUEZ, M J MARTIN, AND J A ABRISQUETA .. 66

A child with a recombinant of chromosome 8 inherited from her carrier mother I C S BARNES, D KUMAR, AND R J M BELL .. 67

'Pure' partial trisomy 2q in a male owing to malsegregation of a maternal translocation t(X;2)(p22·3;q32·1) G PLESSIS, J COUTURIER, C TURLEAU, S DESPOISSES, AND J DELAVENNE .. 70

Partial trisomy 12q24·31→qter E H TAJARA, M VARELLA-GARCIA, AND A C T GUSSON .. 73

Fraser syndrome presenting as monozygotic twins with bilateral renal agenesis G MORTIMER, H P McEWAN, AND J R W YATES .. 76

Familial opposable triphalangeal thumbs associated with duplication of the big toes P MERLOB, M GRUNEBAUM, AND S R REISNER .. 78

No 2 *APRIL* 1985

Editorial .. 81

Frequency and replication status of the fragile X, fra(X)(q27–28), in a pair of monozygotic twins of markedly differing intelligence E TUCKERMAN, T WEBB, AND S E BUNDEY .. 85

First trimester fetal diagnosis of genetic disorders: clinical evaluation of 250 cases B BRAMBATI, G SIMONI, C DANESINO, A OLDRINI, E FERRAZZI, L ROMITTI, G TERZOLI, F ROSSELLA, M FERRARI, AND M FRACCARO .. 92

Cell surface abnormality in clones of skin fibroblasts from a carrier of Duchenne muscular dystrophy J HILLIER, G E JONES, H E STATHAM, J A WITKOWSKI, AND V DUBOWITZ .. 100

A pedigree study of perinatally lethal renal disease A BANKIER, M DE CAMPO, R NEWELL, J G ROGERS, AND D M DANKS .. 104

Necropsy findings in neonatal asphyxiating thoracic dystrophy S B TURKEL, E H DIEHL, AND J A RICHMOND 112

CONTENTS

No 1 *FEBRUARY* 1985

Editorial .. 1

Discordant sex in one of three monozygotic triplets B DALLAPICCOLA, C STOMEO, G FERRANTI, A DI LECCI, AND M PURPURA .. 6

The estimation of recurrence risks in monogenic disorders using flanking marker loci R M WINTER .. 12

Investigation of human chromosome polymorphisms by scanning electron microscopy C J HARRISON, E M JACK, T D ALLEN, AND R HARRIS .. 16

The prevalence of translocations in parents of children with regular trisomy 21: a possible inter-chromosomal effect? R H LINDENBAUM, M HULTÉN, A McDERMOTT, AND M SEABRIGHT .. 24

Reduced penetrance in tuberous sclerosis M BARAITSER AND M A PATTON .. 29

Increased frequency of lymphocytic mitotic non-disjunction in recurrent spontaneous aborters R C JUBERG, J KNOPS, AND P N MOWREY .. 32

Profound limb deficiency, thoracic dystrophy, unusual facies, and normal intelligence: a new syndrome S A AL-AWADI, A S TEEBI, T I FARAG, K M NAGUIB, AND M Y EL-KHALIFA .. 36

Trigonocephaly and the Opitz C syndrome C SARGENT, J BURN, M BARAITSER, AND M E PEMBREY .. 39

Short rib-polydactyly syndrome: a single or heterogeneous entity? A re-evaluation prompted by four new cases R BERNSTEIN, J ISDALE, M PINTO, J DU TOIT ZAAIJMAN, AND T JENKINS .. 46

β^+-thalassaemia in the Po river delta region (northern Italy): genotype and β globin synthesis L DEL SENNO, M PIRASTU, R BARBIERI, F BERNARDI, D BUZZONI, G MARCHETTI, C PERROTTA, C VULLO, Y W KAN, AND F CONCONI .. 54

A family with three independent autosomal translocations associated with 7q32→7qter syndrome H N BASS, R S SPARKES, M M LESSNER, M FOX, B PHOENIX, AND J BERNAR .. 59

Case reports

De novo tandem duplication 9p (p12→p24) with normal GALT activity in red cells T MOTEGI, K WATANABE, N NAKAMURA, T HASEGAWA, AND Y YANAGAWA .. 64

A complex balanced rearrangement involving four chromosomes in an azoospermic man M T RODRIGUEZ, M J MARTIN, AND J A ABRISQUETA .. 66

A child with a recombinant of chromosome 8 inherited from her carrier mother I C S BARNES, D KUMAR, AND R J M BELL .. 67

'Pure' partial trisomy 2q in a male owing to malsegregation of a maternal translocation t(X;2)(p22·3;q32·1) G PLESSIS, J COUTURIER, C TURLEAU, S DESPOISSES, AND J DELAVENNE .. 70

Partial trisomy 12q24·31→qter E H TAJARA, M VARELLA-GARCIA, AND A C T GUSSON .. 73

Fraser syndrome presenting as monozygotic twins with bilateral renal agenesis G MORTIMER, H P McEWAN, AND J R W YATES .. 76

Familial opposable triphalangeal thumbs associated with duplication of the big toes P MERLOB, M GRUNEBAUM, AND S R REISNER .. 78

No 2 *APRIL* 1985

Editorial .. 81

Frequency and replication status of the fragile X, fra(X)(q27–28), in a pair of monozygotic twins of markedly differing intelligence E TUCKERMAN, T WEBB, AND S E BUNDEY .. 85

First trimester fetal diagnosis of genetic disorders: clinical evaluation of 250 cases B BRAMBATI, G SIMONI, C DANESINO, A OLDRINI, E FERRAZZI, L ROMITTI, G TERZOLI, F ROSSELLA, M FERRARI, AND M FRACCARO .. 92

Cell surface abnormality in clones of skin fibroblasts from a carrier of Duchenne muscular dystrophy J HILLIER, G E JONES, H E STATHAM, J A WITKOWSKI, AND V DUBOWITZ .. 100

A pedigree study of perinatally lethal renal disease A BANKIER, M DE CAMPO, R NEWELL, J G ROGERS, AND D M DANKS .. 104

Necropsy findings in neonatal asphyxiating thoracic dystrophy S B TURKEL, E H DIEHL, AND J A RICHMOND 112

An autosomal dominant syndrome with 'acromegaloid' features and thickened oral mucosa H E HUGHES, P J McALPINE, D W COX, AND S PHILIPPS .. 119
Kabuki make-up syndrome (Niikawa-Kuroki syndrome) associated with congenital heart disease S OHDO, H MADOKORO, T SONODA, T NISHIGUCHI, K KAWAGUCHI, AND K HAYAKAWA .. 126
The frequency of lactase phenotypes in Aymara children E BALANZA AND G TABOADA .. 128
The clinical features of the Cohen syndrome: further case reports C NORTH, M A PATTON, M BARAITSER, AND R M WINTER .. 131
Dermatoglyphs in children with mitral valve prolapse J S H TAY, W C L YIP, H K YAP, B W LEE, H B WONG, AND S O CHAY .. 135
Abstracts of the meeting of the Clinical Genetics Society held on 8, 9, and 10 November 1984 at the Royal Free Hospital, London .. 137
Case reports
De novo paracentric inversion in an X chromosome H M HERR, S J HORTON, AND C I SCOTT JR .. 140
Monosomy 13q32·3→qter: report of two cases H RIVERA, S A GONZÁLEZ-FLORES, F RIVAS, J SÁNCHEZ-CORONA, M MOLLER, AND J M CANTÚ .. 142
Congenital diaphragmatic hernia in half sibs A H LIPSON AND G WILLIAMS .. 145
A case of suspected teratogenic holoprosencephaly M STABILE, A BIANCO, S IANNUZZI, M C BUONOCORE, AND V VENTRUTO .. 147
Tracheo-oesophageal anomalies in the Goldenhar anomalad A MENDELBERG, I ARIEL, P MOGLE, AND I ARAD .. 149
Severe pseudoachondroplasia with parental consanguinity I D YOUNG AND J R MOORE .. 150
Short reports
A case of malignant spinal cord ependymoma in association with a duplication of part of the long arm of chromosome 12 B G R NEVILLE, A C BERRY, AND Y STODDART .. 154
Interstitial deletion of chromosome 2 S MARKOVIĆ, M KRSTIĆ, V ŠULOVIĆ, Z RADOJKOVIĆ, AND S ADŽIĆ 154
Familial occurrence of a pseudodicentric chromosome 21 I HANCKE AND K MILLER .. 155
Extra euchromatic band in the qh region of chromosome 9 Z DOCHERTY AND M A HULTÉN .. 156
Book reviews .. 158
Announcements .. 160

No 3 *JUNE* 1985

Editorial .. 161
Review article: Erroneous theories of sex determination U MITTWOCH .. 164
Hypothesis: An hypothesis regarding the origin of aneuploidy in man: indirect evidence from an experimental model M H KAUFMAN .. 171
Linkage analysis of a DNA polymorphism proximal to the Duchenne and Becker muscular dystrophy loci on the short arm of the X chromosome C S BROWN, P L PEARSON, N S T THOMAS, M SARFARAZI, P S HARPER, AND D J SHAW .. 179
Polymorphism of DNA sequence in the human pro α2(I) collagen gene A F GROBLER-RABIE, D K BREBNER, S VANDENPLAS, G WALLIS, R DALGLEISH, R E KAUFMAN, A J BESTER, C G P MATHEW, AND C D BOYD .. 182
Exclusion of the α1(II) cartilage collagen gene as the mutant locus in type IA osteogenesis imperfecta B SYKES, R SMITH, S VIPOND, C PATERSON, K CHEAH, AND E SOLOMON .. 187
Seckel syndrome: an overdiagnosed syndrome E THOMPSON AND M PEMBREY .. 192
A new brachydactyly syndrome with similarities to Julia Bell types B and E P PITT AND I WILLIAMS .. 202
Clinical and haematological evaluation of β thalassaemia intermedia with increased Hb F and Hb A_2 in heterozygotes: β thalassaemia intermedia I C ALTAY AND A GURGEY .. 205
Clinical and haematological evaluation of β thalassaemia intermedia characterised by unusually low Hb F and increased Hb A_2: β thalassaemia intermedia II A GURGEY, S KAYIN, E KANSU, AND C ALTAY .. 213

Case reports
Extra Yq and partial monosomy 12p due to a Y;12 translocation in a boy with features of the 12p deletion syndrome E ORYE, M CRAEN, G LAUREYS, R VAN COSTER, AND B VAN MELE 222
Interstitial deletion of the long arm of chromosome 11 J M KLEP-DE PATER, H F DE FRANCE, AND J B BIJLSMA 224
Interstitial deletion 2q24·3: case report with high resolution banding J BERNAR, R S SPARKES, AND S ALLENSWORTH 226
Hyperinsulinaemic hypoglycaemia in an infant with mosaic trisomy 13 V S SMITH AND G P GIACOIA 228
Complex cardiac malformation in a case of trisomy 9 T WILLIAMS, I ZARDAWI, R QUAIFE, AND I D YOUNG 230
Phenotypic delineation of ring chromosome 15 and Russell-Silver syndromes G N WILSON, S E SAUDER, M BUSH, AND I Z BEITINS 233
Book reviews 237

No 4 *AUGUST* 1985

Editorial 241
Review article: Molecular genetics of the human X chromosome K E DAVIES 243
A population study of adult onset limb-girdle muscular dystrophy J R W YATES AND A E H EMERY .. 250
A community study of severe mental retardation in the West Midlands and the importance of the fragile X chromosome in its aetiology S BUNDEY, T P WEBB, A THAKE, AND J TODD 258
Hereditary sideroblastic anaemia and ataxia: an X linked recessive disorder R A PAGON, T D BIRD, J C DETTER, AND I PIERCE 267
The inheritance of primary lymphoedema R F DALE 274
Zygosity determination in newborn twins using DNA variants C DEROM, E BAKKER, R VLIETINCK, R DEROM, H VAN DEN BERGHE, M THIERY, AND P PEARSON 279
A familial pericentric inversion of chromosome 22 with a recombinant subject illustrating a 'pure' partial monosomy syndrome J L WATT, I A OLSON, A W JOHNSTON, H S ROSS, D A COUZIN, AND G S STEPHEN 283
Survey on haemoglobin variants, β thalassaemia, glucose-6-phosphate dehydrogenase deficiency, and haptoglobin types in Turks from Western Thrace M AKSOY, A KUTLAR, F KUTLAR, G DINÇOL, S ERDEM, AND S BAŞTESBIHÇI 288
Admission of Hb S heterozygotes to a general hospital is relatively reduced in malarial areas B COLOMBO AND L FELICETTI 291
Evaluation of haematological findings in 50 Bahraini patients with sickle cell disease and in some of their parents M A BUHAZZA, A B BIKHAZI, AND F P KHOURI 293
Cornelia de Lange syndrome in several members of the same family D KUMAR, C E BLANK, AND B L GRIFFITHS 296
High incidence of Meckel's syndrome in Gujarati Indians I D YOUNG, A B RICKETT, AND M CLARKE .. 301
Gene deletion in an Italian haemophilia B subject F BERNARDI, L DEL SENNO, R BARBIERI, D BUZZONI, R GAMBARI, G MARCHETTI, F CONCONI, F PANICUCCI, M POSITANO, AND S PITRUZZELLO 305
Two brothers with Martsolf's syndrome J M SÁNCHEZ, C BARREIRO, AND H FREILIJ 308
Familial pulmonary valve stenosis, atrial septal defect, and unique electrocardiogram abnormalities A A CIUFFO, E CUNNINGHAM, AND T A TRAILL 311
Case reports
Complex translocation in a boy with trichorhinophalangeal syndrome J M SÁNCHEZ, J D LABARTA, T C DE NEGROTTI, AND A M MIGLIORINI 314
Interstitial deletion of chromosome 7p detected antenatally K MARKS, L HILL, R G CHITHAM, AND W L WHITEHOUSE 316
Correspondence 319
Book reviews 324
Announcements 328

No 5 *OCTOBER* 1985

Review article: The human Y chromosome P GOODFELLOW, S DARLING, AND J WOLFE 329
Annotation: Classroom teaching in genetics and birth defects: the Nottingham experience J S FITZSIMMONS 345
Chronic proximal spinal muscular atrophy of childhood and adolescence: problems of classification and genetic counselling I HAUSMANOWA-PETRUSEWICZ, J ZAREMBA, AND J BORKOWSKA 350
Genetics and biochemical variability of variants of 21 hydroxylase deficiency M T GORDON, D I CONWAY, D C ANDERSON, AND R HARRIS 354
The genetic control of phenformin 4-hydroxylation R R SHAH, D A P EVANS, N S OATES, J R IDLE, AND R L SMITH 361
A family study of hidradenitis suppurativa J S FITZSIMMONS AND P R GUILBERT 367
Osmotic fragility test in heterozygotes for α and β thalassaemia L MACCIONI AND A CAO 374
The first observation of Hb D Punjab β^o thalassaemia in an English family with 22 cases of unsuspected β^o thalassaemia minor among its members S WORTHINGTON AND H LEHMANN 377
Phenotypic variation in LADD syndrome E THOMPSON, M PEMBREY, AND J M GRAHAM 382
Three new cases of oculodentodigital (ODD) syndrome: development of the facial phenotype M A PATTON AND K M LAURENCE 386
Familial absence of the pectoralis major, serratus anterior, and latissimus dorsi muscles T J DAVID AND R M WINTER 390
Abstracts of the meeting of the Clinical Genetics Society held on 24 and 25 April 1985 at the University of Birmingham 393
Case reports
Reciprocal translocation between chromosomes 8 and 9 in atypical chronic myeloid leukaemia D G OSCIER, G J MUFTI, A GARDINER, AND T J HAMBLIN 398
Partial 2p deletion in a girl with a complex chromosome rearrangement involving chromosomes 2, 6, 11, and 21 R S YOUNG, M A MEDRANO, AND K L HANSEN 401
Sacrococcygeal teratoma and normal alphafetoprotein concentration in amniotic fluid M SZABÓ, P VARGA, A ZALATNAI, J HIDVÉGI, Z TÓTH, AND Z PAPP 405
The Nager acrofacial dysostosis syndrome with the tetralogy of Fallot E THOMPSON, R CADBURY, AND M BARAITSER 408
Correspondence 411
Obituary 416

No 6 *DECEMBER* 1985

Editorial 417
Sir Cyril Clarke 418
Biographical details of contributors to special issue 419
Prenatal diagnosis of the common haemoglobin disorders D J WEATHERALL, J M OLD, S L THEIN, J S WAINSCOAT, AND J B CLEGG 422
Marcella O'Grady Boveri (1865–1950) and the chromosome theory of cancer V A MCKUSICK 431
Application of an intragenic genomic probe to genetic counselling for haemophilia B in the west of Scotland J M CONNOR, A F PETTIGREW, I M HANN, C D FORBES, G D O LOWE, AND N A AFFARA 441
Genetic linkage between Huntington's disease and the DNA polymorphism G8 in South Wales families P S HARPER, S YOUNGMAN, M A ANDERSON, M SARFARAZI, O QUARRELL, R TANZI, D SHAW, P WALLACE, P M CONNEALLY, AND J F GUSELLA 447
Immunogenetic studies in families with rheumatoid arthritis and autoimmune thyroid disease P A SANDERS, D M GRENNAN, P A DYER, G G DE LANGE, AND R HARRIS 451
Should women at high risk of neural tube defect have an amniocentesis? K M LAURENCE, G ELDER, K T EVANS, B M HIBBARD, M HOOLE, AND C J ROBERTS 457
Prenatal diagnosis of ornithine carbamoyl transferase deficiency using a gene specific probe M E PEMBREY, J M OLD, J V LEONARD, C H RODECK, R WARREN, AND K E DAVIES 462

Collagen genes and proteins in osteogenesis imperfecta F M POPE, A C NICHOLLS, J McPHEAT, P TALMUD, AND R OWEN 466
The acetylator phenotypes of Saudi Arabian diabetics D A PRICE EVANS, S PATERSON, P FRANCISCO, AND GALVAREZ 479
Familial complex autosomal translocations involving chromosomes 7, 8, and 9 exhibiting male and female transmission with segregation and recombination S WALKER, P J HOWARD, AND D HUNTER 484
HLA antigens in psoriasis and psoriatic arthritis J C WOODROW AND A ILCHYSYN 492
Book reviews 496
Index 505